What is Maple Syrup Urine Disease?

Maple Syrup Urine Disease (MSUD) is an inherited metabolic disorder that, if untreated, causes mental retardation, physical disabilities and death. First described as a disease in 1954, it is a rare disorder, believed to be in all ethnic groups worldwide. The national incidence is 1 in 225,000 births.

MSUD derives its name from the sweet, burnt sugar, or maple syrup smell of the urine. The disorder affects the way the body metabolizes (processes) certain components of protein. These components are the three branched-chain amino acids leucine, isoleucine, and valine. These amino acids accumulate in the blood causing a toxic effect that interferes with brain functions.

How do you get Maple Syrup Urine Disease?

MSUD is a recessive genetic disease. This means both parents of a child with MSUD must carry a mutation in the same gene (the same genetic code). These mutated genes do not function normally, thus causing disease. A person with a gene mutation is called a carrier. Carriers are not affected by their one abnormal gene. (Just as everyone carries genetic codes, such as those for the color of hair and eyes, everyone also carries several abnormal genes, which can cause disease.)

Each individual person has two genes that code for the enzyme activity involved in the breakdown of the branched-chain amino acids —one gene coming from the father and one from the mother. If one gene functions normally, but the other does not, then the person is a carrier of MSUD. If neither gene functions normally, then that person has MSUD. It is also possible to receive the normal gene from each parent and not have any mutated genes for the disease.

When parents are both carriers of the gene for MSUD, with *each pregnancy,* there is a 1 in 4 chance of having a baby with MSUD, a 2 in 4 chance that the baby will have only one gene for MSUD and be a carrier like the parents, and a 1 in 4 chance that the baby will neither be a carrier nor have MSUD.

Each parent of a child with MSUD carries a defective gene for MSUD along with a normal gene. The defective gene is a recessive gene, therefore

parents are called "carriers" and are not affected by the disease. Each child receives one gene from each parent.

When both parents are carriers, there is a 1 in 4 chance with each pregnancy that the baby will receive a defective gene from each parent and have MSUD; a 2 in 4 chance the baby will receive one defective and one normal gene becoming a carrier of MSUD; and a 1 in 4 chance the baby will receive two normal genes. Persons with two normal genes cannot pass MSUD to their offspring.

How common is Maple Syrup Urine Disease?

The national incidence is 1 in 225,000 births.

How is Maple Syrup Urine Disease treated?

Initially the infant will be given an MSUD formula supplemented with carefully controlled amounts of a protein-based formula such as Enfamil or Similac. The protein-based formula provides the infant with the limited amount of branched-chain amino acids needed to grow and develop normally. Frequent monitoring of blood levels and adjustments in the formula mixture are necessary to ensure the correct balance of leucine, isoleucine, and valine.

As children with MSUD grow, they continue taking the special formula. They are allowed other foods which are weighed or measured in the home to supply the prescribed amount of leucine each day. Typically the MSUD diet does not include any high protein foods such as meat, nuts, eggs, and most dairy products. Children gradually learn to accept the responsibility for controlling their diets. There is no age at which diet treatment can be stopped. Lifelong therapy is essential for an optimal outcome.

This information is adapted in part from the
MSUD Family Support Group

The Missouri Department of Health and Senior Services provides these links as alternative information to the public health community. If you have any additions or corrections, please contact us.

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Where can I obtain services for Maple Syrup Urine Disease in Missouri?

Metabolic Treatment Centers:

Cardinal Glennon Children's Hospital
1465 South Grand Blvd.
St. Louis, MO 63104
(314) 577-5639

Children's Mercy Hospital
2401 Gillham Road
Kansas City, MO 64108
(816) 234-3290

St. Louis Children's Hospital
One Children's Place
St. Louis, MO 63110
(314) 454-6093

University of Missouri Health Care
One Hospital Drive
Columbia, MO 65212
(573) 882-6991

What services does DHSS provide?

See Metabolic Formula Distribution Program FACT sheet.